

Severe Combined Immunodeficiency (SCID)

Low levels of T-cell Receptor Excision Circles (TRECs) are associated with Severe Combined Immunodeficiency (SCID). Other conditions associated with low TRECs include reticular dysgenesis, coronin-1A deficiency and thymic aplasia/complete DiGeorge syndrome. T lymphocytes fail to develop, and the affected infant may also have impaired B lymphocyte function.

Inheritance: Autosomal recessive and X-linked

Estimated Incidence: 1:40,000 to 1:60,000

Abnormal Screen Result: Decreased TRECs (T-cell receptor excision circles)

Method of Notification: All abnormal results are shared with the provider of record and may be sent to an Immune Disorder Specialist, upon verbal and written request.

Next Steps if Abnormal: **Potential medical emergency when TRECs are very low to absent!**

See infant as soon as possible to ascertain health status. **Consult a pediatric specialist (immunology or pediatric infectious disease)** and initiate diagnostic evaluation and treatment as recommended. Common diagnostic studies include specialized flow cytometry and molecular testing to determine specific mutations.

Report all findings to the SC Newborn Screening Program via fax at 803-898-0337.

In addition, repeat TREC on filter paper and send to the DHEC Public Health Laboratory. Low TRECs with questionable RPP30 (internal lab control) may indicate poor quality sample, DNA amplification failure, anticoagulant interference, or other condition, (aka) inconclusive results. Prompt repeat screening is necessary to rule out SCID in these infants.

Neonatal Presentation: Usually none. Median age for onset of symptoms is 8 weeks of age.

Emergency Treatment: Usually none.

Standard Treatment: Bone marrow transplantation by 3 months of age is associated with the best outcomes for SCID. Infants with other conditions may be treated with medications.

Special Considerations

Infectious Disease Precautions - Parents should be instructed to avoid exposure of the infant to anyone with viral/bacterial illnesses until an immunodeficiency is definitively ruled out. No vaccines should be given until cleared to do so by the specialist.

The specialist may also advise mothers to suspend breastfeeding while their blood is checked for anti-CMV IgG antibodies and CMV DNA. These mothers should be encouraged to pump and freeze their breast milk during this time. Prompt resumption of breastfeeding is encouraged if the mother is seronegative.

Only leukoreduced, CMV negative, irradiated blood should be used if a transfusion is necessary.

Premature/Sick Infants—Premature infants may have low TRECs due to immaturity of the immune system. Prompt repeat screening is indicated. The pediatric specialist (immunology or pediatric infectious disease) may recommend flow cytometry if TRECs are low in a second blood spot specimen.

NOTE: Low TRECs may also be found in specimens obtained from infants who have undergone thymectomy/cardiac surgery if the specimen is collected after surgery.